Table 9. *BBS7* Pathologic Allelic Variants

Gene	Mutation	Exon	Reference
BBS7	p.H323R homozygous	10	Badano et al 2003
BBS7	p.H323R homozygous	10	Badano et al 2003
BBS7	p.T2111 homozygous	6	Badano et al 2003
BBS7	K237fsX296 homozygous	7	Badano et al 2003

.0001 BBS7 H323R. This amino acid substitution was identified in the homozygous state in all affected individuals from two unrelated BBS pedigrees [Badano et al 2003].

.0002 BBS7 T211I. This amino acid substitution was identified in the homozygous state in all individuals in a consanguineous BBS pedigree [Badano et al 2003a]. All affected family members additionally carried a E234K heterozygous change in the *BBS1* gene, raising the possibility of complex inheritance between BBS7 and BBS1 [Badano et al 2003].

.0003 BBS7 K237fsX296. A 4 base pair deletion within exon 7of the BBS7 gene, resulting in the introduction in a premature stop codon within exon 9, was identified in the homozygous state in the only affected individual in a BBS family from Saudi Arabia.